Movement Disorders
CLINICAL PRACTICE
CASE REPORT

Huntington's Disease Revealed by Familial Cervical Dystonia

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Mrs. M (a 45-year-old woman) had progressively developed isolated cervical dystonia (CD) over a 12-month period. She had no history of long-term treatment with medications in general or antipsychotics in particular. With the exception of CD, the results of a neurological examination (including psychiatric and cognitive status) were completely normal. Mrs. M did not report a family history of neurological disorders at the time of our initial interview but had said that she was no longer in touch with family members as a result of a dispute, raising the possibility that her knowledge of her family's medical history might have been inaccurate or biased. However, Mrs. M was then lost to follow-up. Five years later, she consulted for memory problems (although CD was still the predominant clinical sign). A neuropsychological assessment revealed mild cognitive impairment (memory impairment, constructional apraxia, and cognitive dysexecutive syndrome), and a clinical assessment evidenced mild, generalized chorea. In the time since her first consultation, Mrs. M had re-established a relationship with her family and now reported a family history of movement disorders (suggesting the inheritance of an autosomal-dominant condition) (Fig. 1). Taken as a whole, these data prompted us to diagnose Huntington's disease (HD), which was subsequently

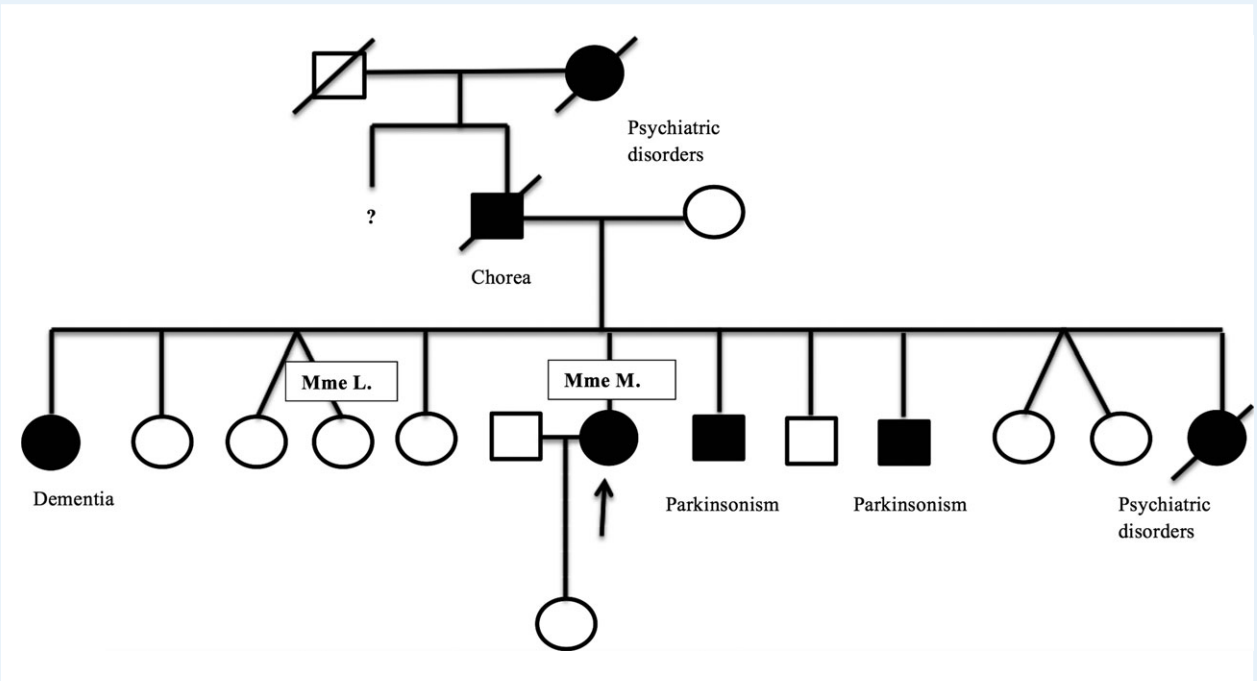

Figure 1 Mrs M's family tree.

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confirmed by a positive cytosine-adenine-guanine (CAG) expansion test (n=45 repeats) for the huntingtin gene (*HTT*) allele.

Two years later, Mrs. L (a 49-year-old woman) was admitted for recent, mild postural instability; in fact, her most prominent clinical feature was CD. There was no evidence of cognitive decline, psychiatric disorders, or chorea. The CD had started 7 years earlier and had remained an isolated disorder until the appearance of postural instability. Given the absence of significant disability, the patient had not previously consulted a neurologist. Furthermore, she reported that a sister of hers also suffered from CD. We then realized that our first patient (Mrs. M) was Mrs. L's sister. Mrs. L also tested positive for *HTT* CAG expansion (n = 44). Currently, CD is still the main clinical feature in both sisters (Fig. 2. Video 1; see online supporting information).

CD is an abnormal, involuntary movement disorder that occurs in about 14% of patients with HD.[1] Although CD is often 1 of the initial symptoms of juvenile HD,[2] it is considered to be atypical in the early stages of adult-onset HD.[3] The first case of adult-onset HD revealed by CD was reported by Ashizawa and Jankovic[4] but has since been subject to debate; Lang[5] reported a case of isolated CD in a patient with a documented familial history of HD but normal genetic test results and dismissed Ashizawa's association as a coincidence (because CD is the most common form of adult-onset focal dystonia). In the 2 present cases, CD was the first manifestation of adult-onset HD, and both sisters displayed a very similar clinical picture.

Figure 2 Mrs L. The cervical dystonia predominates; there are no choreic movements.

Here, we reported on the first cases of isolated familial focal CD revealing HD. One could argue that CD in the 2 sisters might be due to coincidental dystonia 7 (DYT7), DYT1, DYT6, or DYT25 primary torsion dystonia; however, this is very unlikely because focal dystonia (and especially focal CD) has hardly ever been reported in the context of DYT1 or DYT6; and, to our knowledge, the association between DYT7 or DYT25 and HD has not been reported.[6,7]

In conclusion, HD should be considered as a potential cause of isolated familial CD.

Author Roles

1. Research Project: A. Conception, B. Organization, C. Execution; 2. Statistical Analysis: A. Design, B. Execution, C. Review and Critique; 3. Manuscript Preparation: A. Writing the First Draft, B. Review and Critique.

D.A.: 3A
M.T.:3B
O.G.: 3B
P.K.: 3B

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Supporting Information

A video accompanying this article is available in the supporting information here.

Video 1: Mme M. The cervical dystonia is still the predominant clinical problem, whereas the choreic movements and the cognitive disorders are still very mild.

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